



hypomyelination and congenital cataract

Hypomyelination and congenital cataract is an inherited condition that affects the nervous system and the eyes. This disease is one of a group of genetic disorders called leukoencephalopathies. Leukoencephalopathies involve abnormalities of the brain's white matter. White matter consists of nerve fibers covered by a fatty substance called myelin. Myelin insulates nerve fibers and promotes the rapid transmission of nerve impulses. Hypomyelination and congenital cataract is caused by a reduced ability to form myelin (hypomyelination). Additionally, people with this disorder are typically born with a clouding of the lens (cataract) in both eyes.

People with this condition usually have normal development throughout the first year of life. Development slows around the age of 1. Most affected children learn to walk between the ages of 1 and 2, although they usually need some type of support. Over time they experience muscle weakness and wasting (atrophy) in their legs, and many affected people eventually require wheelchair assistance. Weakness in the muscles of the trunk and a progressive abnormal curvature of the spine (scoliosis) further impair walking in some individuals. Most people with hypomyelination and congenital cataract have reduced sensation in their arms and legs (peripheral neuropathy). In addition, affected individuals typically have speech difficulties (dysarthria) and mild to moderate intellectual disability.

Frequency

The prevalence of hypomyelination and congenital cataract is unknown.

Genetic Changes

Mutations in the *FAM126A* gene cause hypomyelination and congenital cataract. The *FAM126A* gene provides instructions for making a protein called hyccin, the function of which is not completely understood. Based on the features of hypomyelination and congenital cataract, researchers presume that hyccin is involved in the formation of myelin throughout the nervous system. Hyccin is also active in the lens of the eye, the heart, and the kidneys. It is unclear how mutations in the *FAM126A* gene cause cataracts.

Most *FAM126A* gene mutations that cause hypomyelination and congenital cataract prevent the production of hyccin. People who cannot produce any hyccin have problems forming myelin, leading to the signs and symptoms of this condition.

People who have mutations that allow some protein production tend to have milder symptoms than those who produce no protein. These individuals typically retain the

ability to walk longer, although they still need support, and they usually do not have peripheral neuropathy.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- HCC

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Hypomyelination and Congenital Cataract
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C2674508/>

Other Diagnosis and Management Resources

- GeneReview: Hypomyelination and Congenital Cataract
<https://www.ncbi.nlm.nih.gov/books/NBK2587>
- MedlinePlus Encyclopedia: Congenital Cataract
<https://medlineplus.gov/ency/article/001615.htm>
- MedlinePlus Encyclopedia: Muscle Atrophy
<https://medlineplus.gov/ency/article/003188.htm>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Encyclopedia: Congenital Cataract
<https://medlineplus.gov/ency/article/001615.htm>
- Encyclopedia: Muscle Atrophy
<https://medlineplus.gov/ency/article/003188.htm>
- Health Topic: Leukodystrophies
<https://medlineplus.gov/leukodystrophies.html>
- Health Topic: Peripheral Nerve Disorders
<https://medlineplus.gov/peripheralnervedisorders.html>
- Health Topic: Scoliosis
<https://medlineplus.gov/scoliosis.html>

Genetic and Rare Diseases Information Center

- Hypomyelination and congenital cataract
<https://rarediseases.info.nih.gov/diseases/11980/hypomyelination-and-congenital-cataract>

Additional NIH Resources

- National Eye Institute: Facts About Cataract
https://nei.nih.gov/health/cataract/cataract_facts
- National Institute of Neurological Disorders and Stroke: Peripheral Neuropathy Fact Sheet
<https://www.ninds.nih.gov/Disorders/All-Disorders/Peripheral-Neuropathy-Information-Page>

Educational Resources

- Boston Children's Hospital: Cataracts in Children
<http://www.childrenshospital.org/conditions-and-treatments/conditions/cataracts>
- Boston Children's Hospital: Nervous System Disorders in Children
<http://www.childrenshospital.org/conditions-and-treatments/conditions/n/nervous-system-disorders>
- Cleveland Clinic: Cataracts in Children
<http://my.clevelandclinic.org/childrens-hospital/health-info/diseases-conditions/hic-cataracts-in-children>
- Cleveland Clinic: Neuropathy
<http://my.clevelandclinic.org/health/articles/neuropathy>

- Disease InfoSearch: Hypomyelination and Congenital Cataract
<http://www.diseaseinfosearch.org/Hypomyelination+and+Congenital+Cataract/8617>
- MalaCards: hypomyelination and congenital cataract
http://www.malacards.org/card/hypomyelination_and_congenital_cataract
- Merck Manual Home Edition for Patients and Caregivers: Cataract
<http://www.merckmanuals.com/home/eye-disorders/cataract/cataract>
- Orphanet: Hypomyelination-congenital cataract syndrome
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=85163

Patient Support and Advocacy Resources

- Prevent Blindness America: Cataract
<http://www.preventblindness.org/cataract>
- The Foundation Fighting Blindness
<http://fffb.ca/>
- The Myelin Project
<http://myelin.org/>
- University of Kansas Medical Center Resource List: Developmental Delay/Mental Retardation
<http://www.kumc.edu/gec/support/devdelay.html>

GeneReviews

- Hypomyelination and Congenital Cataract
<https://www.ncbi.nlm.nih.gov/books/NBK2587>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28hypomyelination%5BTIAB%5D%29+AND+%28congenital+cataract%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- LEUKODYSTROPHY, HYPOMYELINATING, 5
<http://omim.org/entry/610532>

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